Chromosome 16p11.2 Copy Number Variants

Sometimes a person is born with chromosomes that are different. One of the person’s chromosomes may have extra genetic material or missing genetic material. When this happens, it is called a copy number variant, or CNV.

A 16p11.2 “duplication” means there is extra genetic material. A 16p11.2 “deletion” means that genetic material is missing. People with a duplication and people with a deletion are likely to have different features and symptoms.

16p11.2 has two distinct regions, the “proximal” and the “distal.” The typical region is where duplications and deletions are most commonly found. The distal region is next to it.

You can find which section of your or your child’s chromosome 16 is duplicated or deleted in your genetics lab report.

**BP4-BP5 DUPICATION:** Extra genetic information appears in the ~29.5-30.1 Mb area. A person with a BP4-BP5 duplication may have speech delay, developmental delay and intellectual disability, autism spectrum disorder, behavioral issues, and small head size. Read more here.

**BP4-BP5 DELETION:** Genetic information is missing from the area of ~29.5-30.1 Mb. A person with a BP4-BP5 deletion may have speech and language disorders, developmental delay and intellectual disability, learning difficulties, autism spectrum disorder, behavioral issues, a tendency toward obesity, large head size, and seizures. Read more here.

**DISTAL DUPICATION:** Extra genetic information appears in the ~28.8-29.1 Mb area. A person with a distal duplication may have developmental delay and intellectual disability, autism spectrum disorder, speech delay, and seizures.

**DISTAL DELETION:** Genetic information is missing from the area of ~28.8-29.1 Mb. A person with a distal deletion may have developmental delay and intellectual disability, speech delay, autism spectrum disorder, behavioral issues, early feeding difficulties, and a tendency toward obesity.

Everyone is unique. People with a 16p11.2 CNV exhibit different features. Even members of the same family may show a wide range of features.

We are still learning about 16p11.2.

With the help of families, we can gain a better understanding through research. Scientists have discovered a lot about 16p 11.2 with the help of families. That being said, we still do not know everything. If you would like to learn more, join our community, or contribute to the ongoing research, please visit simonssearchlight.org.

*Note: Mb is a genetic unit of length*